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An interesting presentation of a sacrococcygel teratoma with an associated neuroendocrine tumor: A case report

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ABSTRACT

Teratomas are the most frequently occurring germ cell tumors, with 45–65 percent occurring in the sacrococcygeal region [1]. The case presented below involves a newborn male with a malignant sacrococcygeal teratoma and a number of congenital deformities including a left lower extremity amputation, an absent left kidney, hypoplastic and absent left hemipelvis musculature. After excision of the teratoma and subsequent chemotherapy, the patient returned with a persistent abnormal soft tissue, containing persistent malignant elements around the left femur, suspicious for a neuroendocrine tumor. This case shows a rare association between two exceedingly uncommon malignancies.

Teratomas are the most frequently occurring germ cell tumors, with 45–65 percent occurring in the sacrococcygeal region [1]. The majority of sacrococcygeal teratomas (SCT's) in neonates are benign [2]. The case presented here involves a newborn male with a malignant sacrococcygeal teratoma and a number of congenital deformities; including a left lower extremity amputation, an absent left kidney, hypoplasia of the left hemipelvis musculature, as well as ambiguous genitalia. After excision of the teratoma and subsequent chemotherapy, the patient returned with persistent abnormal soft tissue containing malignant elements around the left femur. The clinical presentation of this malignant sacrococcygeal teratoma and its possible involvement in the congenital deformities present at birth is a rare finding and one not seen in the current literature.

1. Case report

A newborn male, gestational age 38.6 weeks and weighing 2.86 kg, was prenatally diagnosed with an apparent isolated amniotic band syndrome with amputation of the left lower extremity. At birth, the infant was noted as having significantly more congenital anomalies than originally detected on prenatal ultrasound. There was extensive involvement of the left lower portion of his body including: Rectal prolapse, an absent left lower extremity, a bifid penus, and an absent hemiscrotum. Imaging, including a CT and an MRI, identified an absent left kidney, a malpositioned bladder directed toward the right, an absent left iliac bone, and a deformed, partially absent hemisacrum. With the exception of a small femur, the remainder of the left lower extremity was absent. The musculature of the left hemipelvis, including the gluteus maximus/medius, was profoundly hypoplastic. There was a scrotal sac seen anteriorly above the subcutaneous tissues of the right pelvis containing a single testicle, but no left hemiscrotum or left testicle. The imaging demonstrated the physical exam findings of: rectal prolapse and malformed phallus with perineal hypospadias, and a large area of abnormal tissue around the left limb bud.

After complete imaging was obtained, the infant was taken to the operating room for an exam under anesthesia with a diverting colostomy and resection of the prolapsed rectum. During that time, catheters were inserted into the urethra and rectum (Fig. 1). The colostomy was performed and the prolapsed rectum was resected. The perineum was sutured closed.

After resection, the pathology of the prolapsed rectum demonstrated a malignant teratoma with areas of yolk sac/endodermal sinus tumor as well as areas suggestive of embryonal carcinoma. AFP and beta HCG levels were normal for age. Further work up at this time revealed no evidence of metastatic disease. The patient underwent three rounds of chemotherapy with follow up imaging showing no residual disease.

One month following completion of chemotherapy, the patient presented to the Emergency Department with anasarca, tachypnea, tachycardia, and abnormal weight gain. Work up revealed

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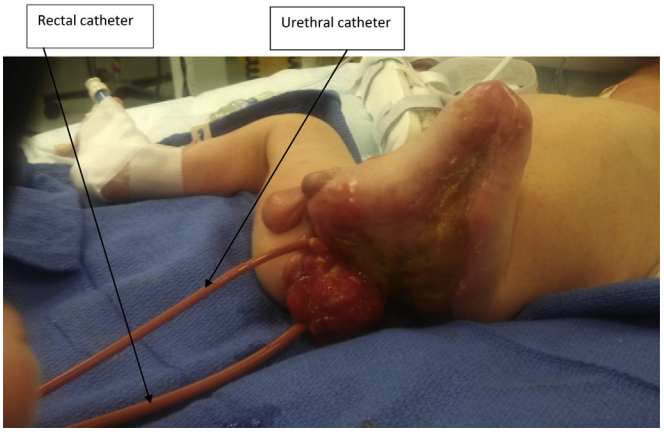

Fig. 1. Intraoperative photo, day of life #5.

hypertension, anemia, thrombocytopenia, and hypoalbuminemia. His severe hypertension and elevated urine metanephrines led to an investigation for a pheochromocytoma or ACTH producing mass. An MRI of his lower extremity was repeated after an MIBG failed to localize a source (Fig. 2). A true cut biopsy of the abnormal appearing tissue along the left femur lower was taken. This revealed small blue cells and ACTH producing cells versus extension and maturation of the original teratoma. All pathology specimens were reviewed and confirmed by an outside pathologist, for this and the previous specimens. A genetic workup undertaken during his hospital course showed a normal karyotype and no known genetic associations.

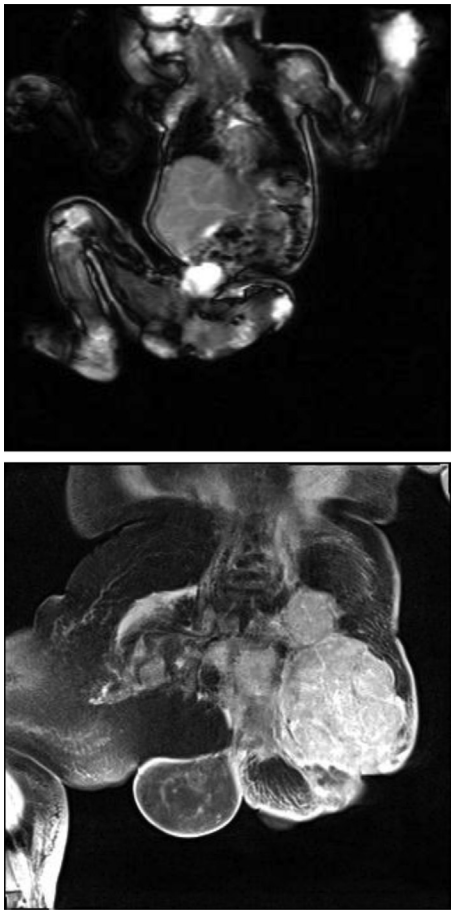

Fig. 2. MRI of the pelvis at 1 day and 5 months showing increase in size of the lesion.

Five days following the muscle biopsy he experienced severe respiratory failure. His parents refused intubation and ultimately withdrew care. Post mortem examination was refused by the parents. He was five months old at the time of his death.

2. Discussion

Extragonadal germ cell tumors, including sacrococcygeal teratomas, are thought to result from abnormal migration and deposition of primordial germ cells [3]. The estimated incidence of sacrococcygeal teratomas is 1 in 27,000 live births [2,4,5]. The classification system for SCTs delineates four distinct types based on anatomic presentation and intra-pelvic involvement, with malignancy rates increasing with each type [6]. A yolk sac component, which produces alpha fetoprotein (AFP), is the most common malignant element [2,7,8]. As a result monitoring AFP levels can be useful in detecting tumor recurrence.

Congenital anomalies are seen in 10–20 percent of cases involving SCTs. Anomalies include tracheoesophageal fistula, imperforate anus, anorectal stenosis, spina bifida, genitourinary malformations, meningomyelocele, and anencephaly [9,10].

Advances in both surgical treatment and the use of chemotherapy have resulted in significant improvement in the overall outcome with germ cell tumors. Studies have reported survival rates ranging from 75% to 90% [11,12]. The standard chemotherapy for malignant extragonadal SCTs includes a combination of cisplatin, etoposide, and bleomycin (PEB) [13–15].

Neuroendocrine tumors such as a pheochromocytomas, have never been reported in the literature in the setting of a malignant SCT. As in our patient, most cases of pheochromocytomas or ACTH producing tumors are diagnosed after histopathology [16]. Pheochromocytomas only make up 3.5% of all malignant neuroendocrine tumors, with patients younger than the age of 5 having a much lower chance of survival [17]. Less than 1% of all pediatric patients, aged 1 month to 18 years, are found to have a pheochromocytoma as the etiology of hypertension. All these patients also had sustained tachycardia, but those in this study did have elevated catecholamines or their metabolites [18]. In the few reported cases of infants presenting with Cushing Syndrome or its features, they have been associated with adrenal tumors, or other neurological tumors [19,20].

Our patient's case was a rare incidence in which a newborn boy presented with a malignant teratoma and shortly thereafter a neuroendocrine tumor. An association between SCTs and the host of congential anomalies seen here, including those to the, limb and pelvic musculature, kidney, and other genitourinary malformations, have not been previously reported in the literature. Additionally, SCT's do not typically present with neuroendocrine components, or in association with a de novo neuroendocrine tumor. Finally, this case was unique in that despite the tumor demonstrating malignant components within the SCTs, yolk sac and embryonal carcinoma, both B-HCG and AFP were normal. This is a case where a patient with multiple phenotypical congenital anomalies was likely the result of a rare presentation of an even rarer combination of malignancies.

Financial disclosure statement

None of the authors have anything to disclose.

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